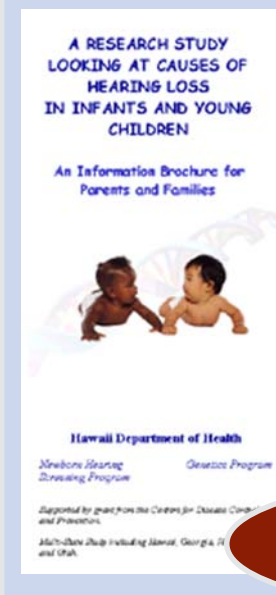


DO YOU KNOW A CHILD WHO IS ELIGIBLE?

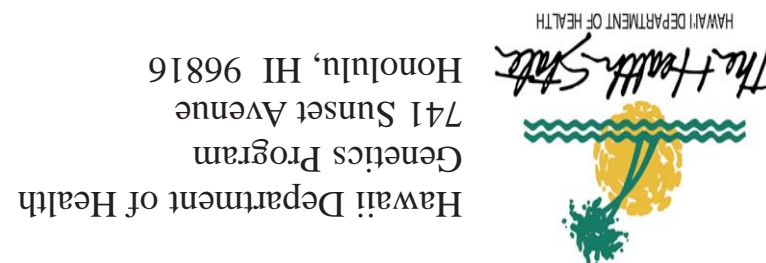
RECRUITMENT FOR HEARING LOSS STUDY IS UNDERWAY...



To refer or find out more,
please contact:

Lianne Hasegawa, MS
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Study Coordinator
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Hawaii Department of Health
Genetics Program
741 Sunset Avenue
Honolulu, HI 96816

Do you know a child who was **born on or after January 1st, 1998** and who was diagnosed with hearing loss before their third birthday?

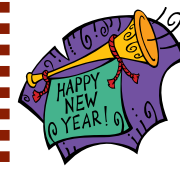
If your answer is YES, this child may be eligible to participate in the Department of Health's research project on the causes of hearing loss in infants and young children.

We are **now accepting referrals** from providers and families. If a child is eligible, the parents will be asked to review and sign a consent form. After the consent has been signed, medical and family history information will be reviewed, and the child's medical records obtained. An initial appointment with a genetic counselor will be scheduled to review this information, details of the study, and what is known about the genetics of hearing loss. Subsequent steps in the study include a visit with a geneticist, and genetic testing (with parental consent). There is no cost to families, and families are under no obligation to participate in this study. The cause of the child's hearing loss may or may not be determined through involvement in this study.

If you are a parent or health care professional and would like more information regarding this study, please contact us. Information for families or providers can be mailed to you.



A Publication of the Hawai'i Department of Health Genetics Program



HAPPY NEW YEAR FROM THE HAWAII GENETICS PROGRAM!!!

The Hawaii Genetics Program and the Newborn Metabolic Screening Program wish all our readers a happy and healthy 2004!

The 2003 year was filled with many exciting developments and milestones in genetics, with DNA Day in April being a monumental event celebrated worldwide. During the past year, many fun and successful activities have been held by the Hawaii Genetics Program across our state, including UH DNA Day, the Genetics For Your Practice conference, various lectures, public health seminar series, focus groups, and participation at many local and national meetings, conferences and events. In addition, we were pleased to report the formation of Hawaii Community Genetics which expands clinical genetics services in Hawaii, and the approval of the Department of Health's Etiology of Congenital Hearing Loss research project. Another much anticipated event in 2003 was the launch of expanded newborn metabolic screening statewide in September.

We anticipate that 2004 will be a year filled with many more exciting developments, events and projects. Throughout the upcoming year, issues of our third volume of Gene News will continue to keep you informed and updated. Happy New Year from all of us!



Coordinator's Corner

The Hawaii Genetics Program is delighted to welcome our newest member, Kirsty McWalter, who joined our team on January 5th, 2004. Kirsty is a 2003 graduate of the Genetic Counseling Program in Cincinnati, Ohio, and is originally from Prince George, Canada. Kirsty was a high school biology teacher prior to entering the world of genetics. Kirsty will play an important role in expanding our program's genetics education activities, and developing educational resources for Hawaii's high schools and other groups across the state. Welcome, Kirsty!

It's taken more time than planned, but we have our new website up and running! We've made the navigation easier and added more information. It will continue as a work in progress since we are still adding additional areas that were requested by website users.

I really like to hear from our readers. If you have comments (good or bad) or questions about GeneNews, our website or any of our activities, please contact me at sylvia@hawaiiigenetics.org or 733-9063. I look forward to hearing from you.

Sylvia M. Au, M.S., C.G.C.
State Genetics Coordinator



Hawaii Genetics & Newborn Metabolic Screening Programs:

Top Row Left to Right :
Sylvia Au, Allison Taylor &
Pauline Mui;

Bottom Row Left to Right:
Loretta Freitas, Christine
Matsumoto, Sharon Hirose,
Lianne Hasegawa & Jan Kong.

Volume 3, Issue 1 January, 2004

Greetings	1
Coordinator's Corner	1
Expanded Newborn Screening	2
Baby Expo	3
Preserving Maleness	3
Recruitment for Hearing Loss	4
Hawaii Community Genetics	Insert



Hawai'i Genetics Program

**Hawai'i Department
of Health**
Children with Special
Health Needs Branch

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Expanded Newborn Screening Pilot Project

What Did We Learn?

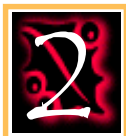


The pilot project results are in the process of being published in peer-reviewed journals.

Update on Statewide Expanded NBS

On September 1st, 2003, Hawaii began mandated NBMS statewide. All infants born in Hawaii now receive expanded NBMS for 31 disorders. Since expanded NBMS is mandated by state law, informed consent is not required. However, informed dissent is required if parents refuse expanded screening for religious reasons.

In support of expanded NBMS, Governor Lingle signed the expanded NBMS revised administrative rules on November 10th, 2003. The new rules, which were reviewed in public hearings and took effect on November 20th, allow the NBMS program to increase the screening fee to \$47 per infant. The fee includes the cost of the expanded screening test, confirmatory testing, and NBMS follow-up. Because expanded NBMS is mandated by law, this fee is covered by insurance.



Although the past year has been hectic for our NBMS program staff, the transition to expanded NBMS has been smooth, and the change has been both rewarding and successful!

During the past year, many exciting events have occurred in the area of newborn metabolic screening (NBMS)!

On August 31st, 2003, Hawaii completed its successful expanded NBMS pilot project, and the results are in! The project, which began on March 1st, 2002, offered expanded NBMS for over 30 disorders to infants born at Kapiolani Medical Center for Women & Children (KMCWC). The project was done in collaboration with the staff of California's NBMS expanded project. The California project was started in January 2002 and ran until June 2003. The results of both projects are shown in the table below:

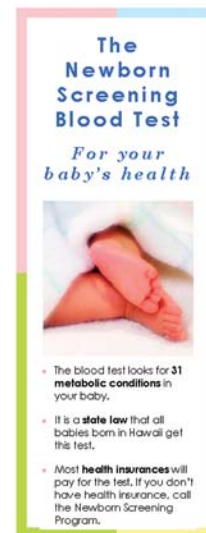
Category	Hawaii # (%)	California # (%)
Number who received expanded NBMS	7,474	354,074
Number with an initial positive result	8 (0.11%)	459 (0.13%)
Number diagnosed with a metabolic disorder	1 (0.01%)	53 (0.01%)

Hawaii's NBMS pilot project also examined the process of informed consent. Informed consent in this project refers to a person voluntarily agreeing to have their baby receive expanded NBMS after reviewing all of the relevant information. In Hawaii's NBMS pilot project, informed consent was obtained by project assistants who spent 15-20 minutes explaining the project to each new mother at KMCWC. Each parent was then asked to complete a satisfaction survey. The survey results show that parents were very satisfied with the individual attention provided by the project assistants, but most did not feel that informed consent was necessary for NBMS. Also, cost analysis revealed that active informed consent is very expensive.

Category	Cost
Cost of informed consent	\$19.64 per family
Total cost of informed consent if applied to all hospitals in Hawaii	\$343,700 per year*

* Amount is equal to almost half of the entire budget of the NBMS program!

PASTEL or PRIMARY?



The New Baby Expo Helps Us Find our True Brochure Colors...

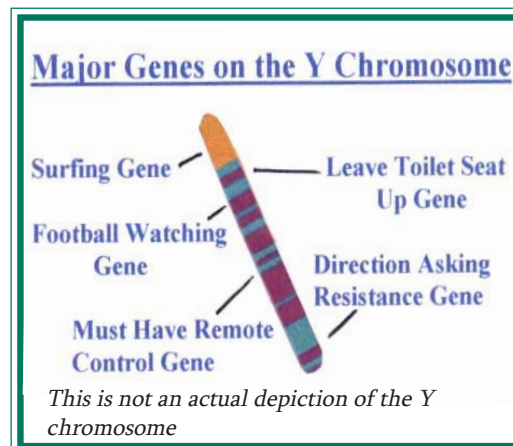


On May 17th and 18th, 2003, Family Expositions held its 6th annual New Baby Expo at the Neil Blaisdell Center. With over 1,000 attendees and close to 100 exhibitors, the expo attracted a variety of people, ranging from soon-to-be parents expecting their first little one to seasoned grandparents with five grandchildren in tow.

The Department of Health's Genetics Program was one of many exhibitors at this busy event, and we took this opportunity to survey the expo attendees about a new newborn metabolic screening (NBMS) brochure. A total of 684 attendees completed the survey.

Most respondents liked pastel colors (56.2%) and the title "The Newborn Screening Blood Test for Your Baby's Health" (45.5%). 80.1% of the attendees preferred the brochure to have a question and answer format, as opposed to a short story. A picture of a baby's hand holding the finger of an adult's hand was the preferred image by (39.0%). In general, respondents preferred pictures of people of the same or similar ethnic background to themselves. This may account for why a picture that is ethnically ambiguous (such as the baby hand holding the adult finger) was more attractive overall.

The majority of respondents believed that the brochure should be easy to understand (81.4%), be informative (63.1%), provide contact information (57.0%), and be simple (55.4%). A smaller number wanted the brochure to be colorful (18.9%) and have many pictures (16.4%). Based on these valuable comments, we are developing a new NBMS brochure that should be released soon. Thank you to everyone who took the time to fill out our surveys!



Preserving Maleness: The Tiny Chromosome Has Lots of Tricks

Human chromosomes come in pairs- one pair being the sex chromosomes- XX in females and XY in males. Each pair of chromosomes swaps and exchanges genetic information as they are copied. Only a very small part of the Y chromosome recombines genetically swaps with the X chromosome; the remaining 95% does not exchange any genetic information. When there is less genetic exchange, there is more accumulation of mutations (genetic changes). As a result of the Y chromosome's lack of genetic exchange,

researchers previously postulated that an accumulation of mutations on this lone chromosome would eventually lead to it shrinking out of existence in the human genome.

Sequencing of the Y chromosome has revealed that it isn't as doomed as previously thought. Since the Y chromosome cannot fully exchange genetic material with another copy of an identical chromosome, it has capitalized on a process called *gene conversion*. The Y chromosome contains back up copies of genes which are used to help fix flaws in gene structure. So, rather than depend on the given paired chromosome like the rest of the human chromosomes do, the Y chromosome does self-repair. Although gene conversion on the Y chromosome is exciting, it is not a perfect process: Mistakes can lead to deletions (missing stretches) of DNA. These deletions may be responsible for a significant amount (at least 10%) of male infertility. On the plus side, specific knowledge of the genetic sequence of the fascinating Y chromosome may better guide diagnosis and treatment in such cases.

Please visit our website at www.hawaii-genetics.org